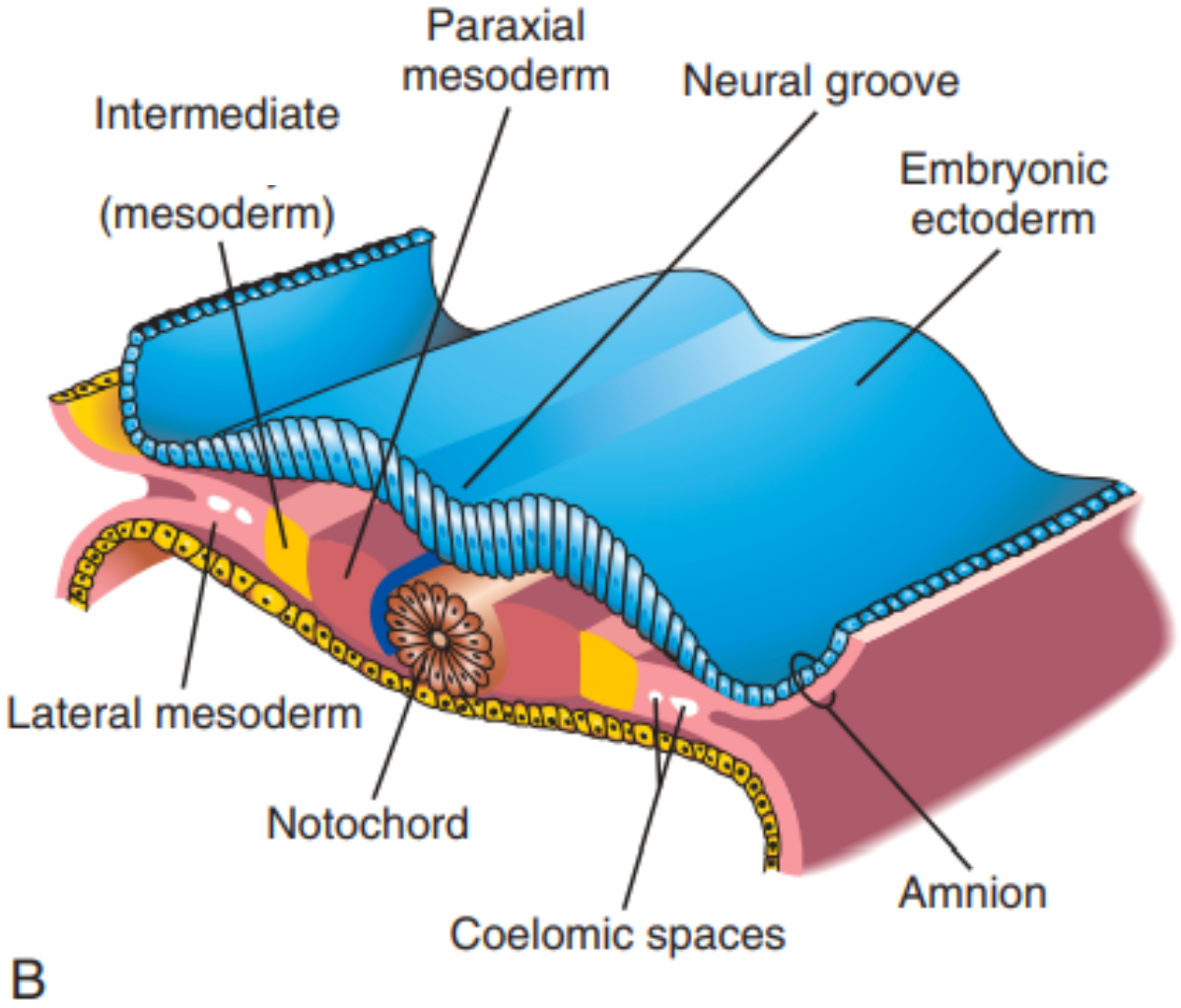
Development and teratology of the urogenital system

25.3.2024

Anna Mac Gillavry

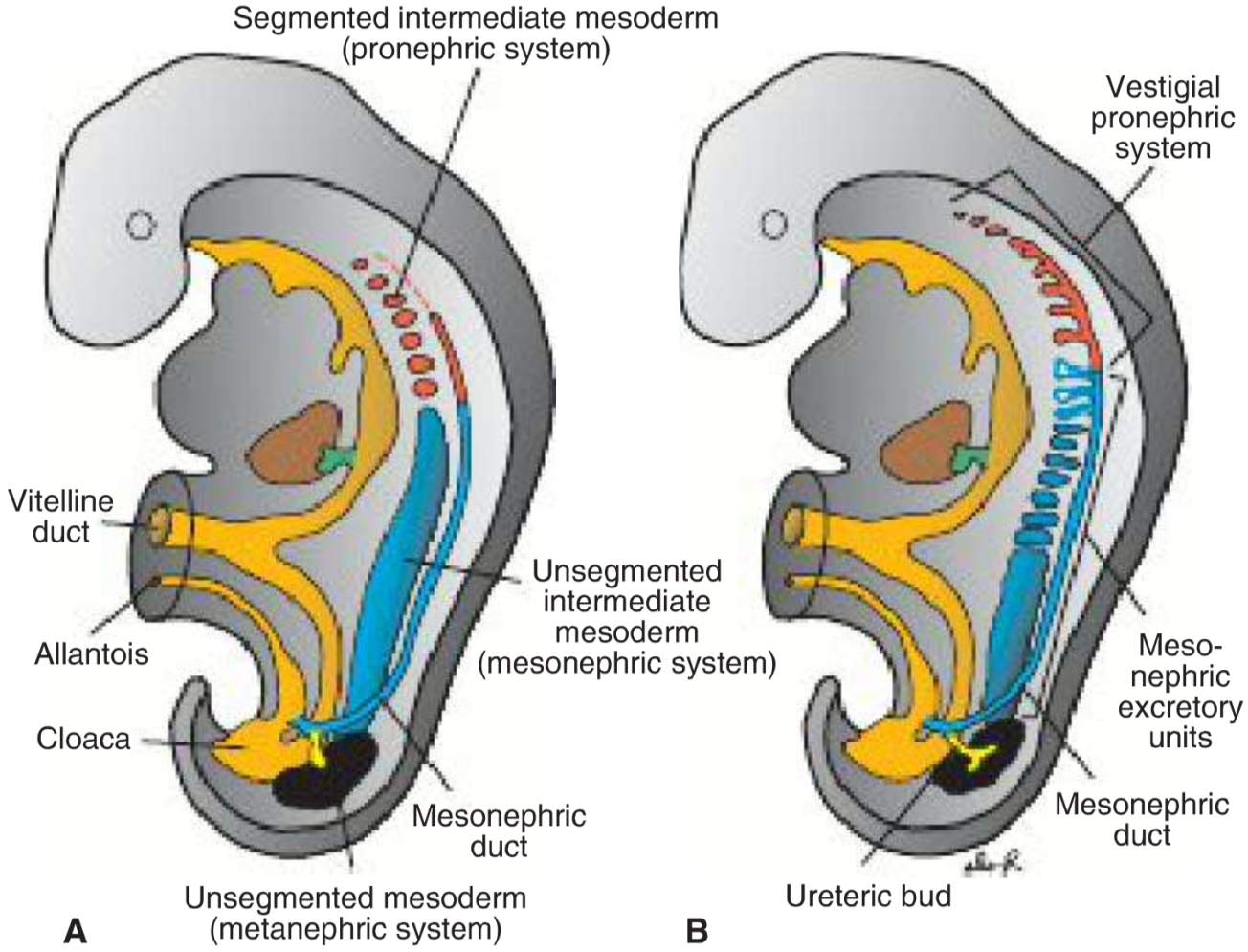
Urinary system



Pronephros: 4th week

Mesonephros: 4th – 10th week (6th – 12th week)

Metanephros: 5th to 9th week (12th week)



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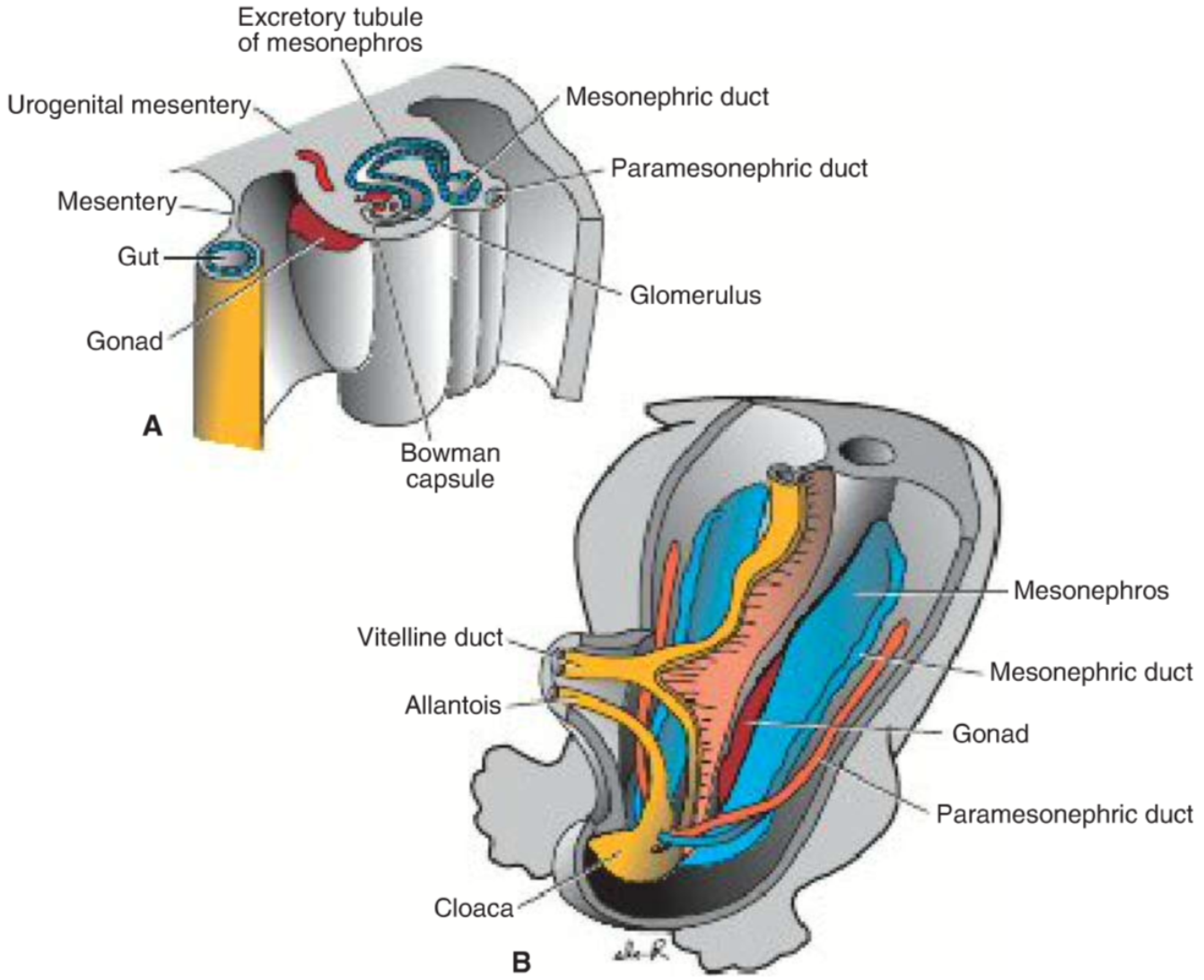
Mesonephros



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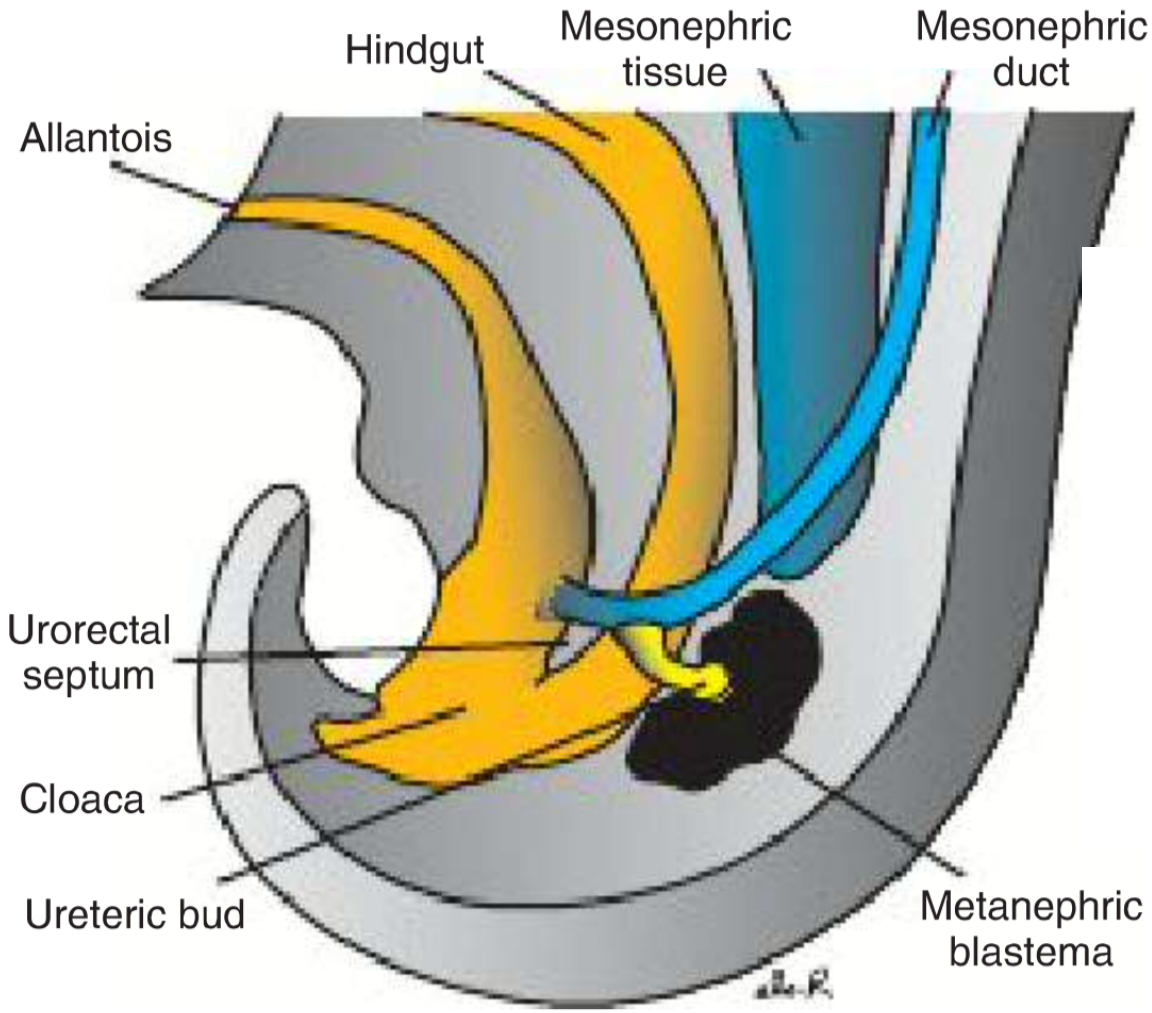
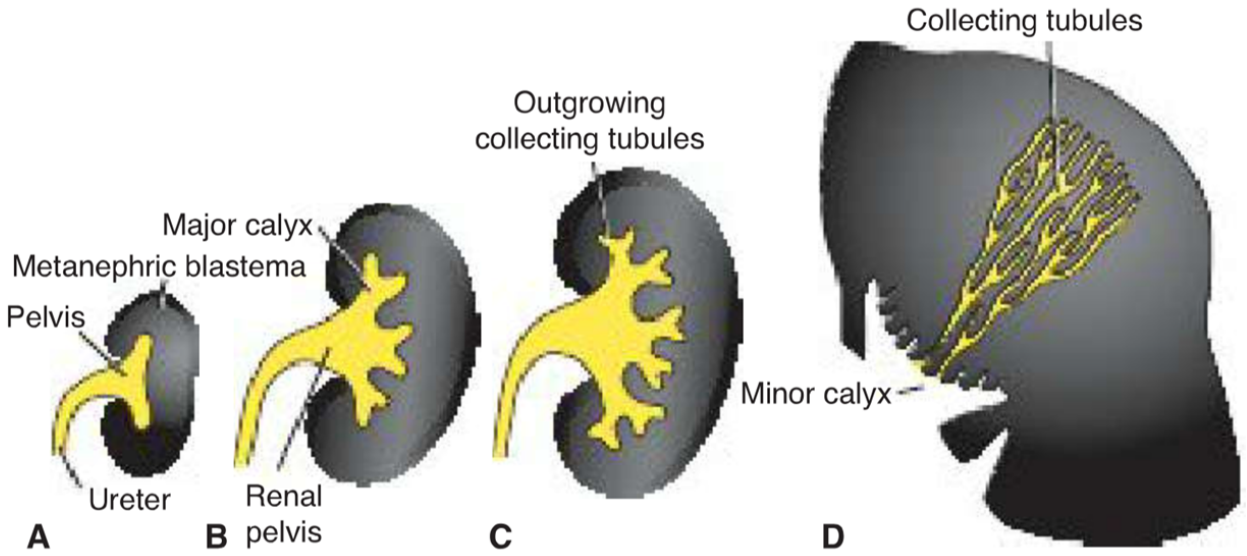
Metanephros





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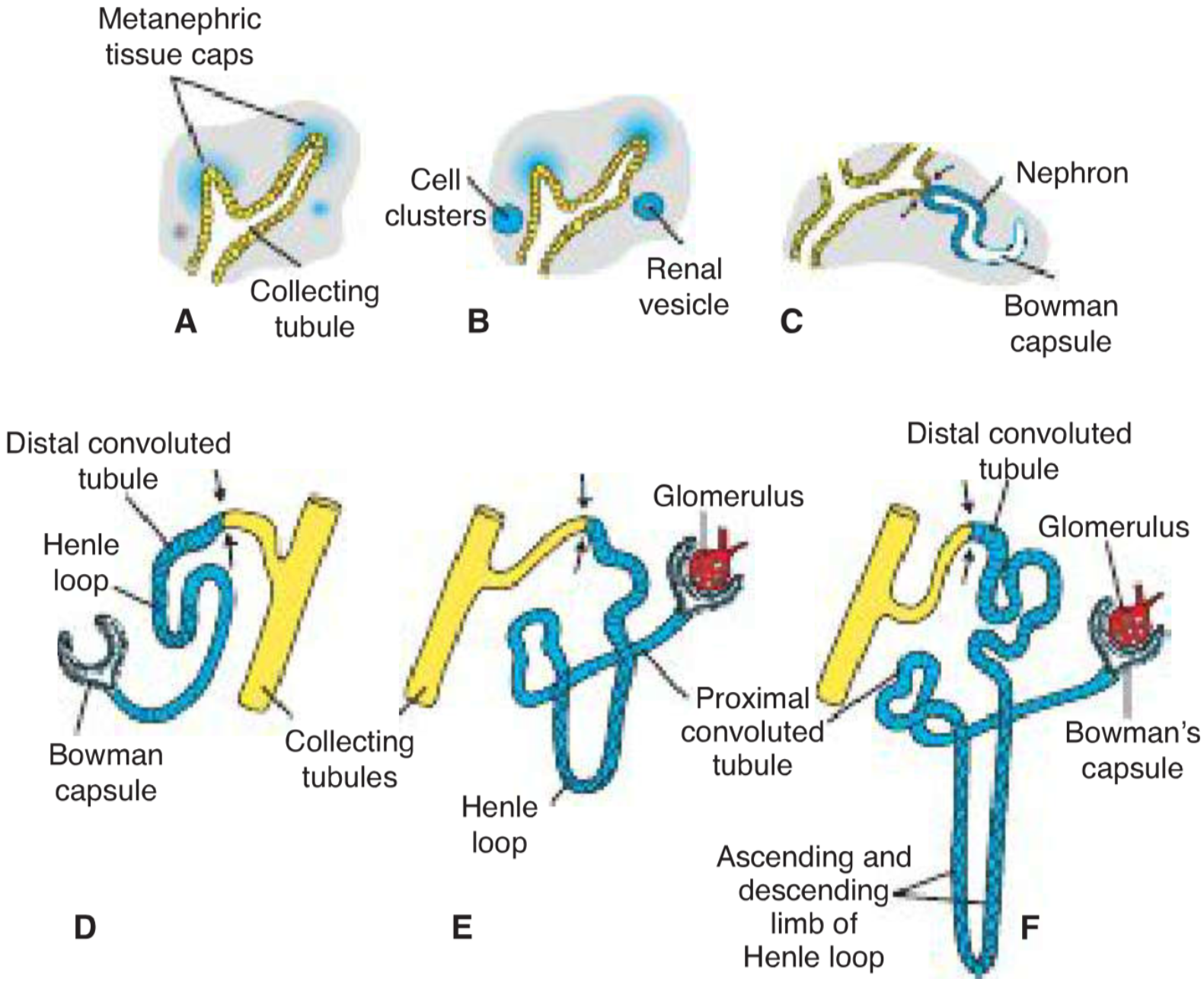

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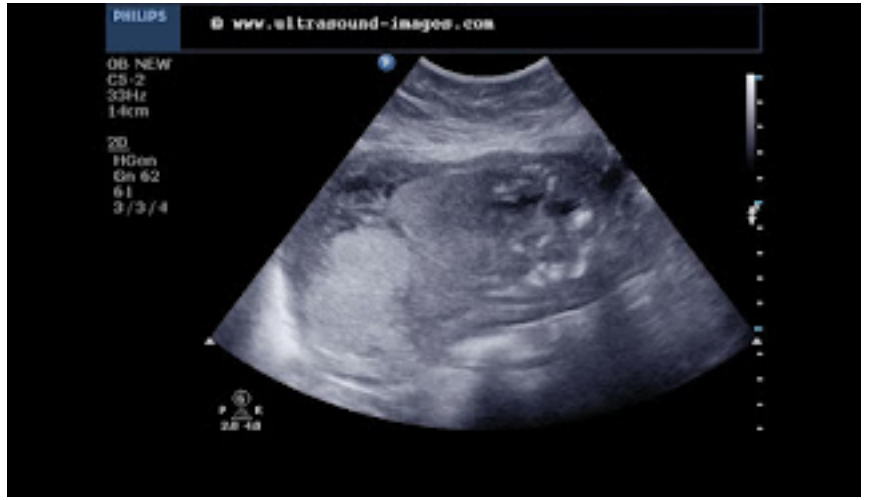
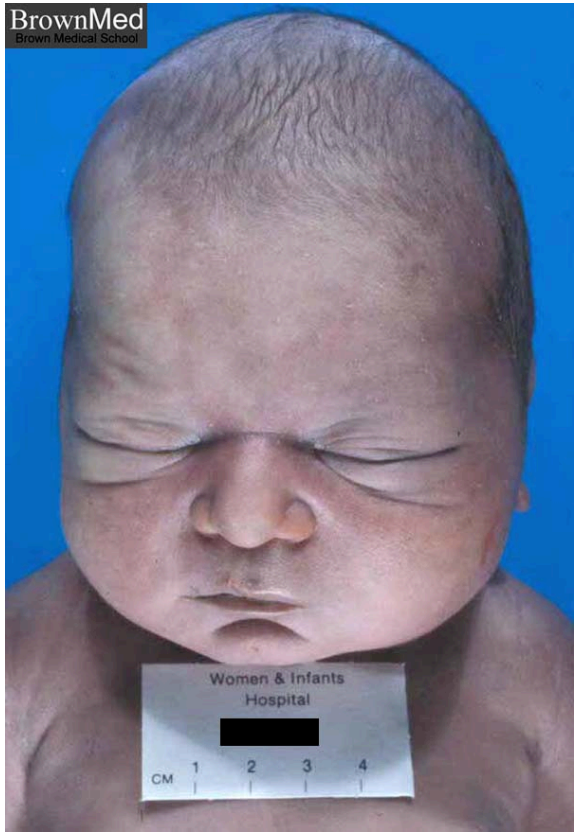
Renal defects





https://www.brown.edu/Courses/Digital_Path/systemic_path/renal/R18

- Renal agenesis:

- unilateral (1/1000);
- bilateral (1/3000 – 10000);
- 3:1 males to females;
- Potter sequence: anuria, oligohydramnios, pulmonary hypoplasia, Potter face

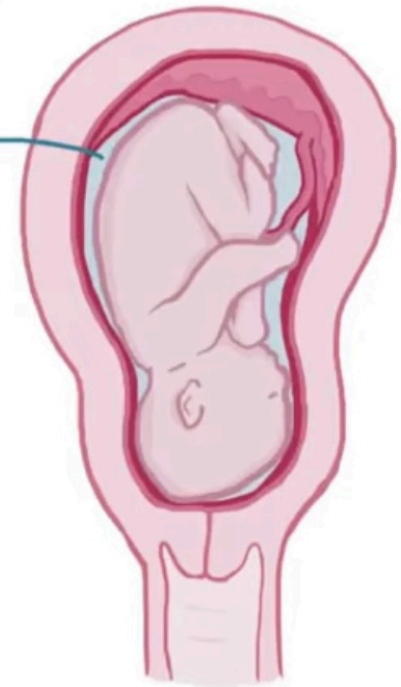
https://1.bp.blogspot.com/-CW1NqZyu_Sk/UIrM1XyuUaI/AAAAAAAADZk/KdZn-NisJ0M/s320/oligohydramnios-severe-1eA.jpg

POTTER SEQUENCE

* (MOSTLY) AFFECTS NEWBORN MALES *

* CAUSED BY OLIGOHYDRAMNIOS *

LOW AMNIOTIC FLUID



PULMONARY HYPOPLASIA

OLIGOHYDRAMNIOS

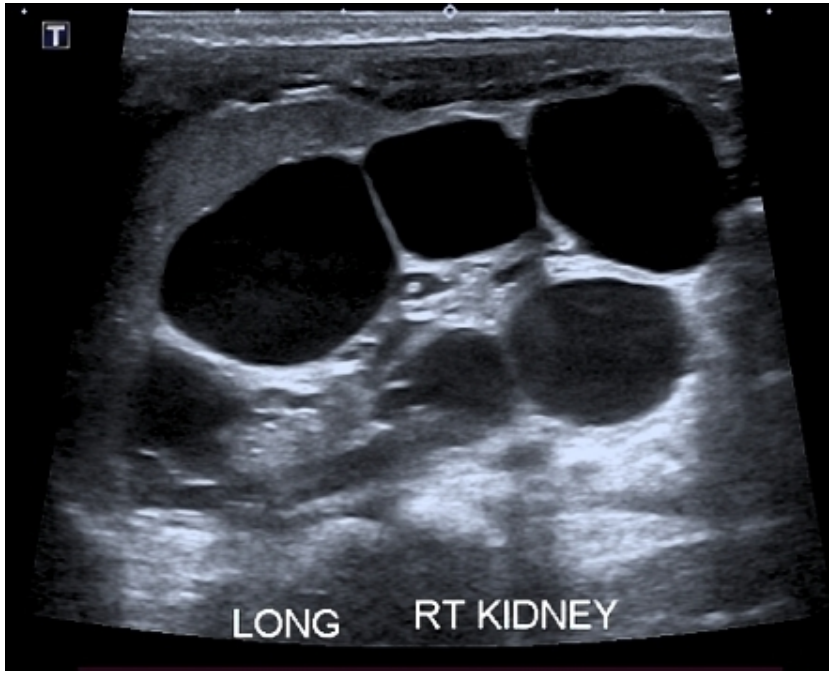
TWISTED SKIN (WRINKLES)

TWISTED FACE

EXTREMITY DEFORMITIES

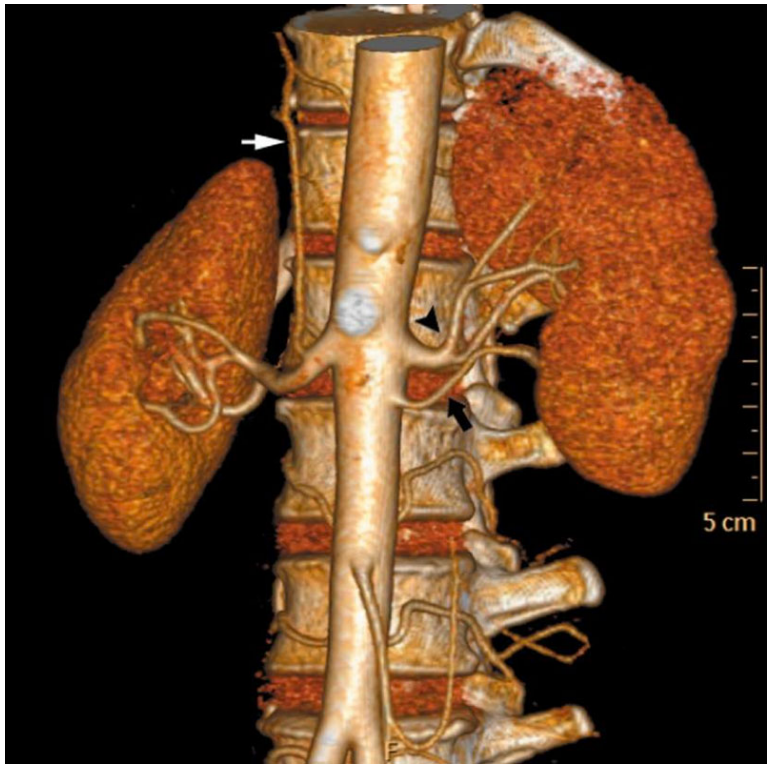
RENAL AGENESIS

- Renal displasia
  - Multicystic displastic kidney
- Congenital polycystic kidney disease:
- autosomal recessive (1/5000)
- autosomal dominant (1/500-1000)
  - group of cilliopathties Bardet-Biedl syndrome, Mackel-Gruber syndrom (lethal)



https://prod-images-static.radiopaedia.org/images/54791799/B_gallery.jpeg

- Duplication of the ureter – splitting of the uretric bud
- Ectopic ureter – development of two uretric buds
- Supernumerary kidney



Abnormal location:

- pelvic kidney
- horseshoe kidney (1/600)
- unilatelar fused kidney
- accessory (suprenumeral) renal arteries –

25 % of kidneys have 2 to 4 arteries

https://embryology.med.unsw.edu.au/embryology/images/b/b8/Accessory_renal_artery.jpg

Renal tumors

- Wilms tumor – affects children by 5 years of age incl. fetal period – WAGR syndrom (microdeletion on chromsome 11 *WT1* and *PAX6* genes)

  Wilms tumor

  Aniridia

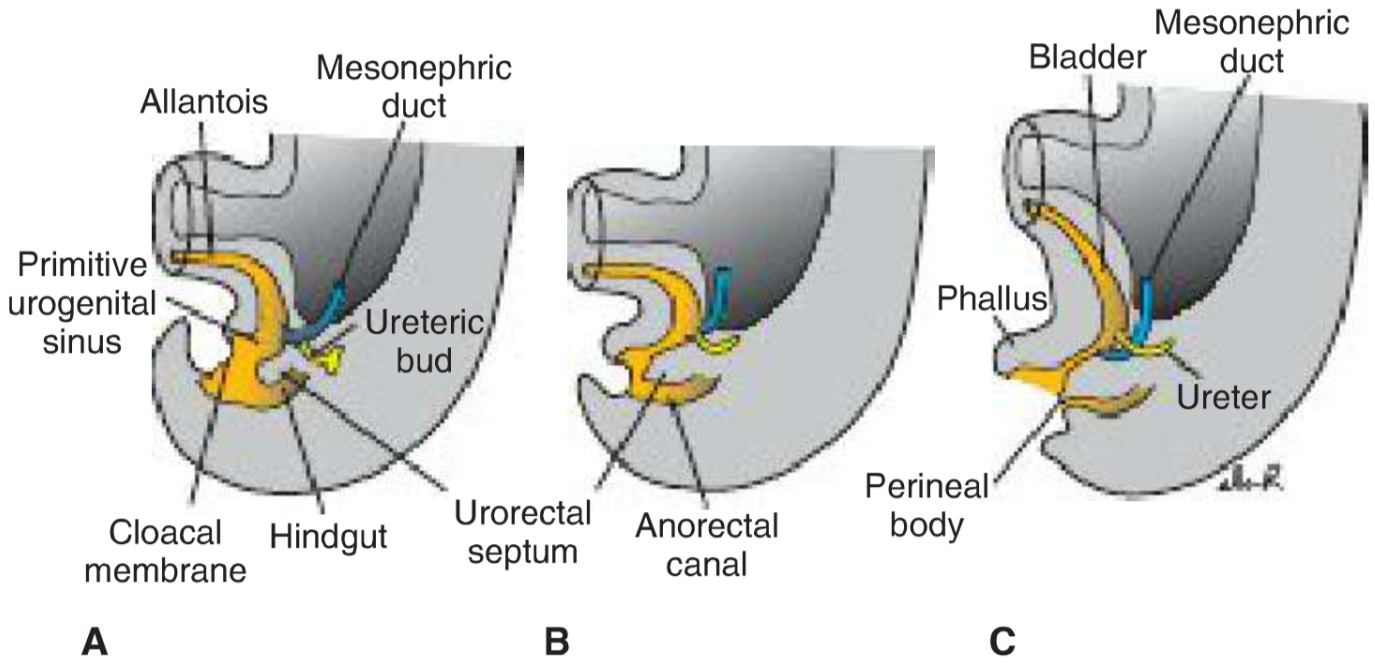
  Gonadoblastomas

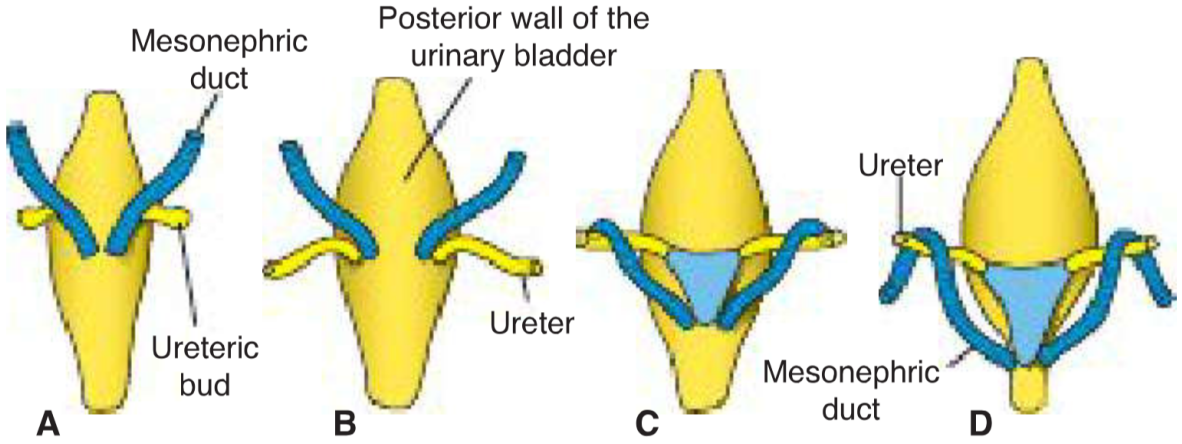
  Retardation (intellectual disability)

  Denys-Drash syndrom

Bladder and urethra

Urogenital sinus:
- vesical part
- pelvic part
- phallic part





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Urachal abnormalities:

- urachal fistula
- urachal cyst
- urachal sinus

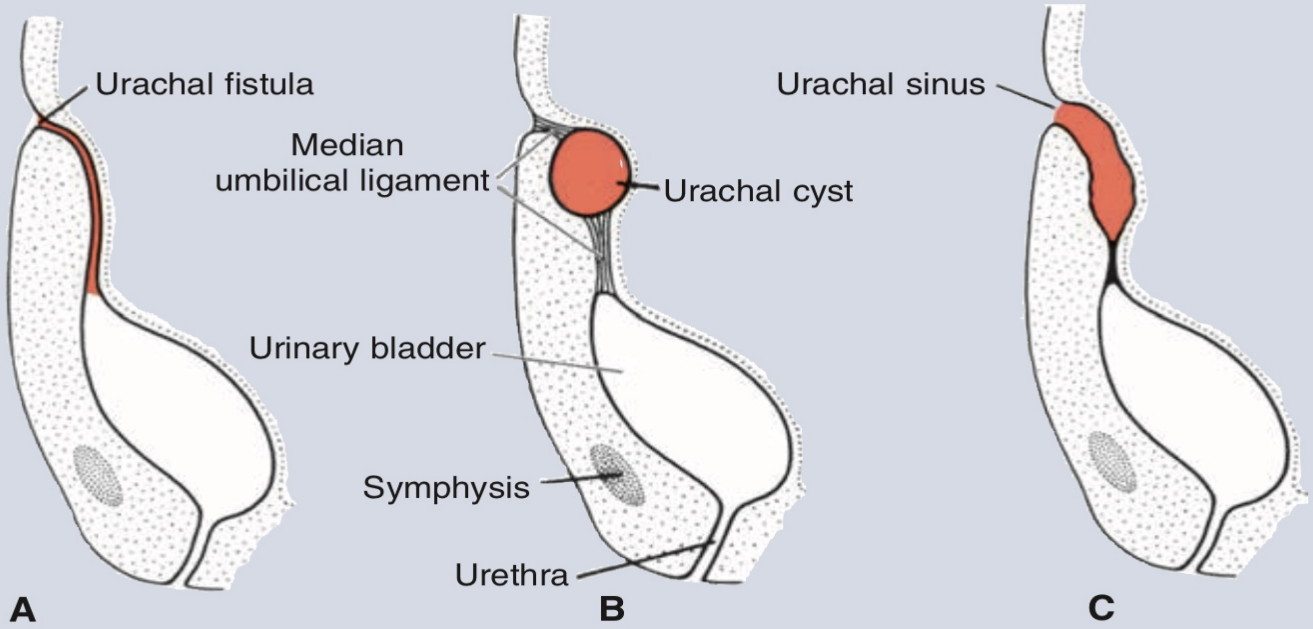

Figure 16.15 **A.** Urachal fistula. **B.** Urachal cyst. **C.** Urachal sinus. The sinus may or may not be in open communication with the urinary bladder.

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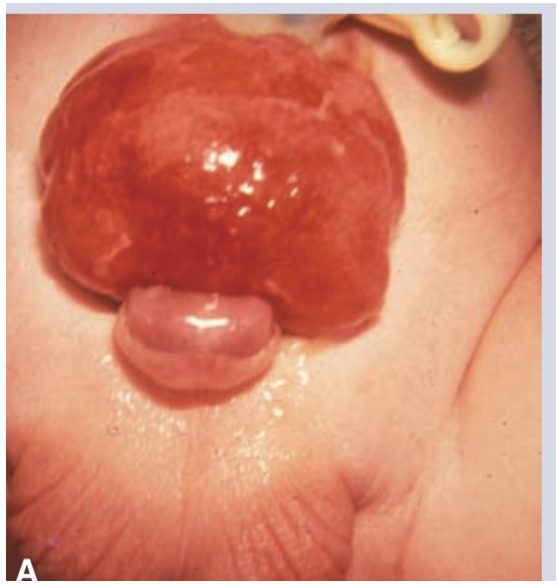
Ventral body wall deffects

- Extrophy of the bladder: (1/50000)
- Extrophy of the cloaca: (1/200000)



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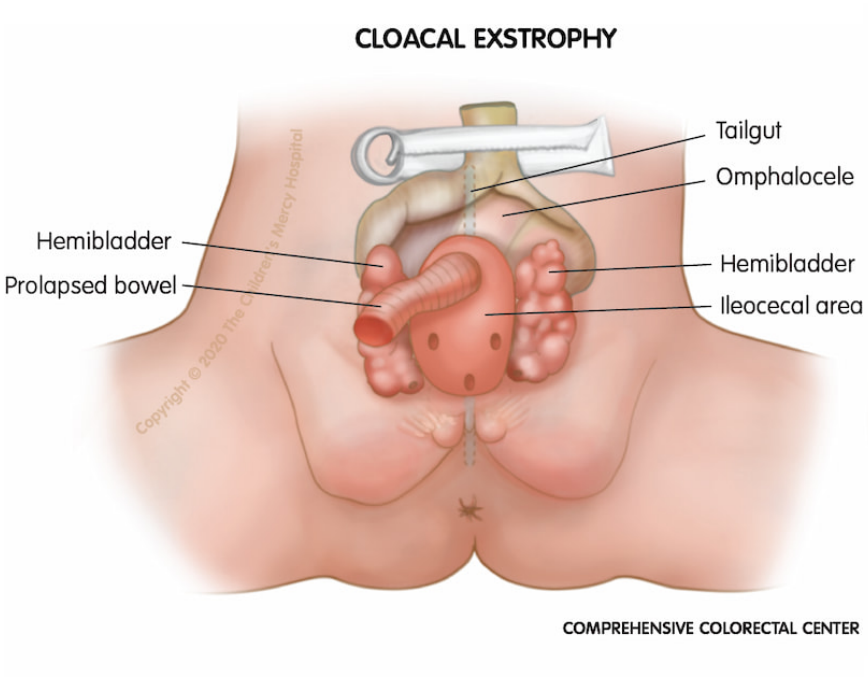
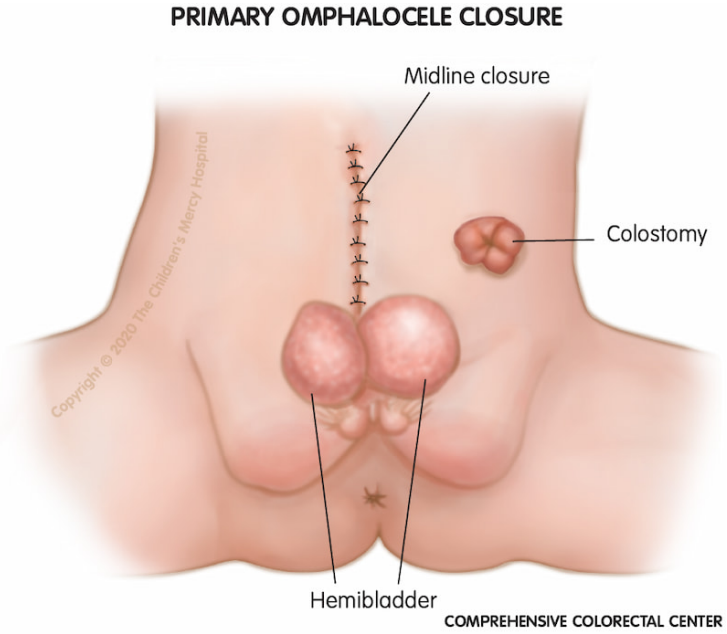
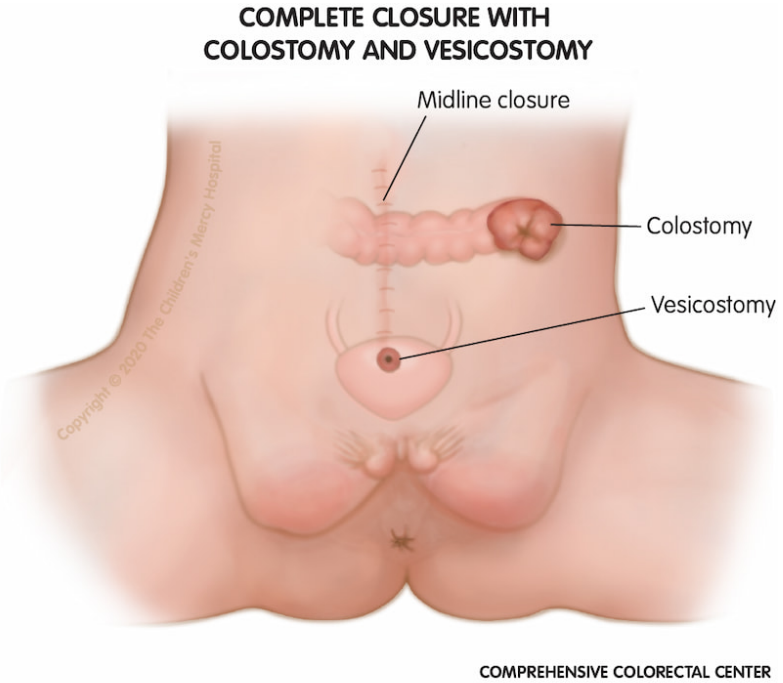
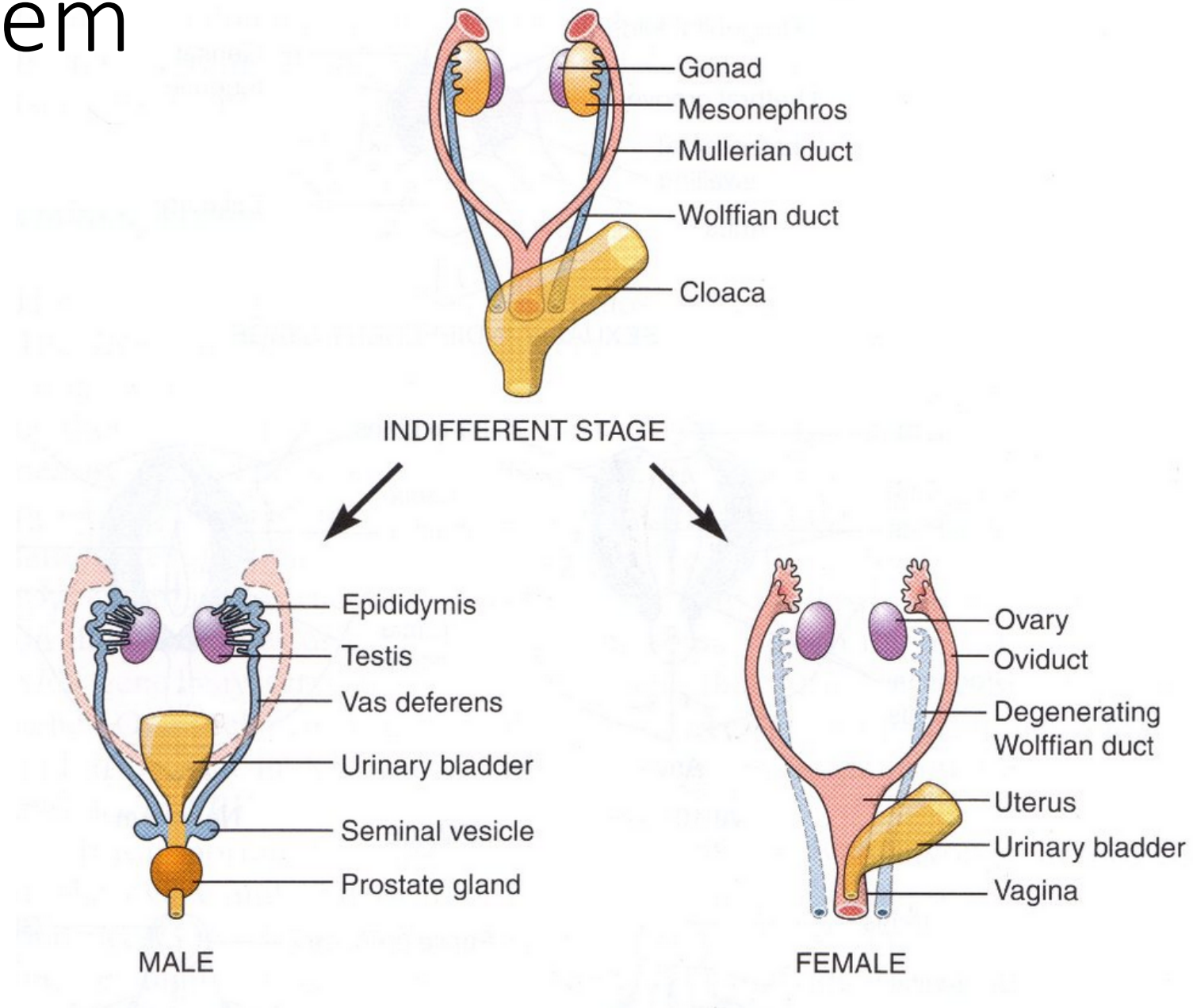






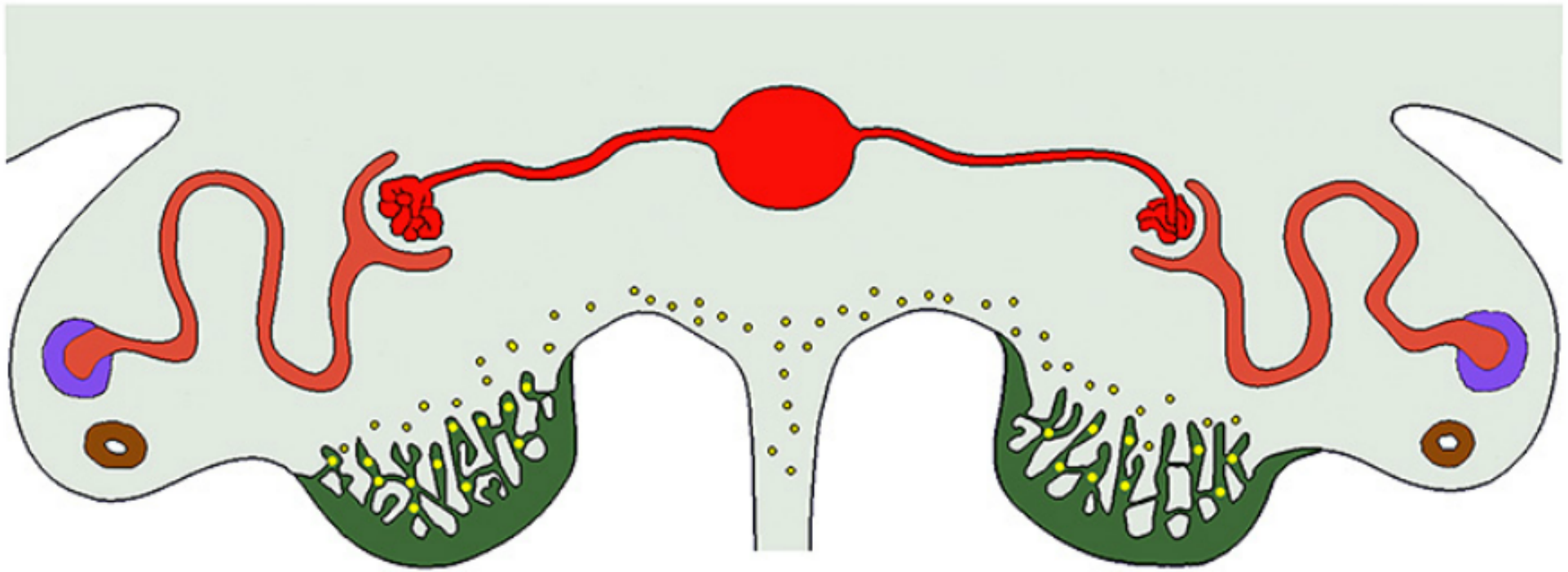
https://www.childrensmercy.org/departments-and-clinics/colorectal-center/anorectal-malformation/cloacal-exstrophy/

Reproductive system

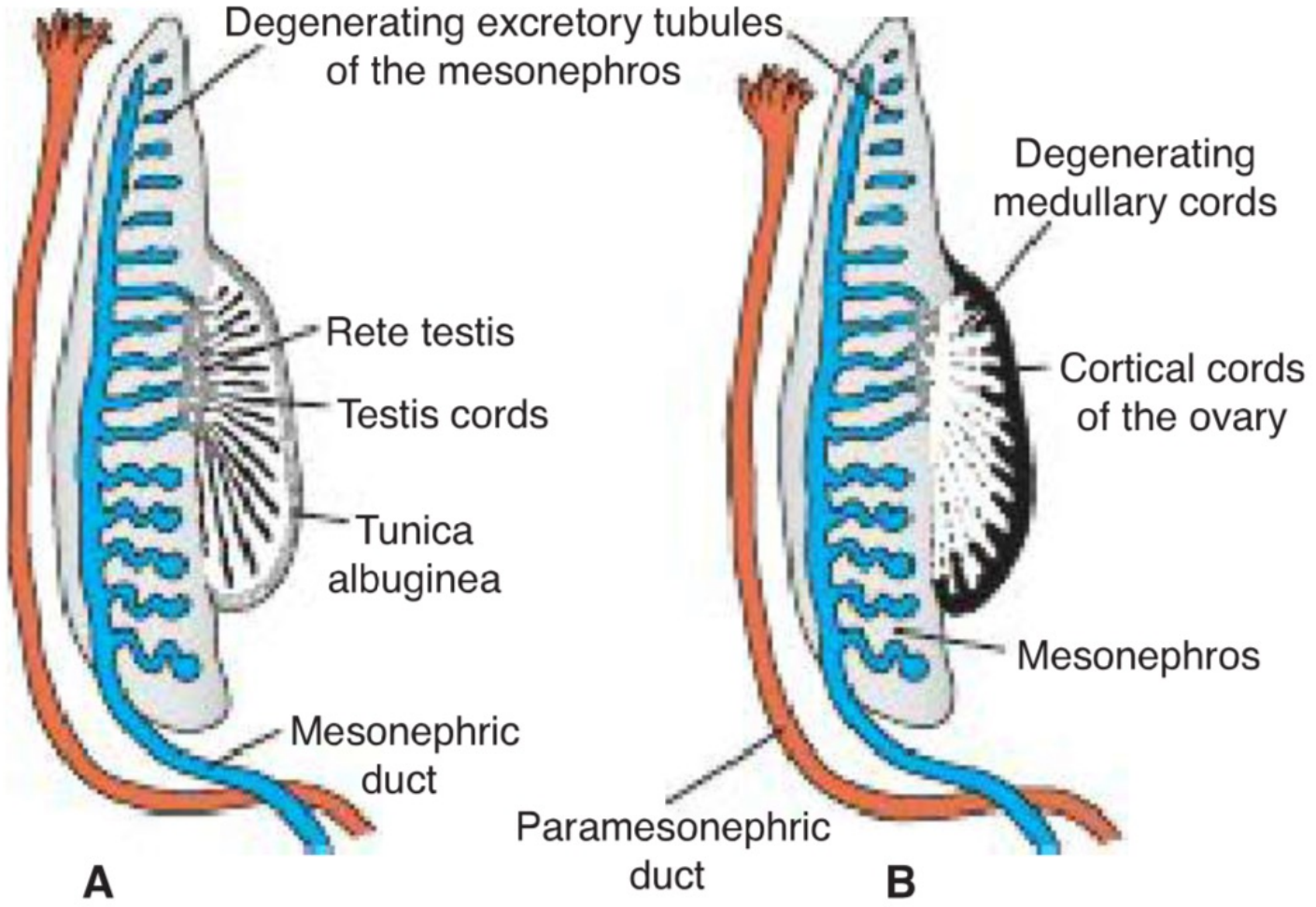


www.dailykos.com

Indifferent stage of the gonads



Indifferent stage of the genital ducts



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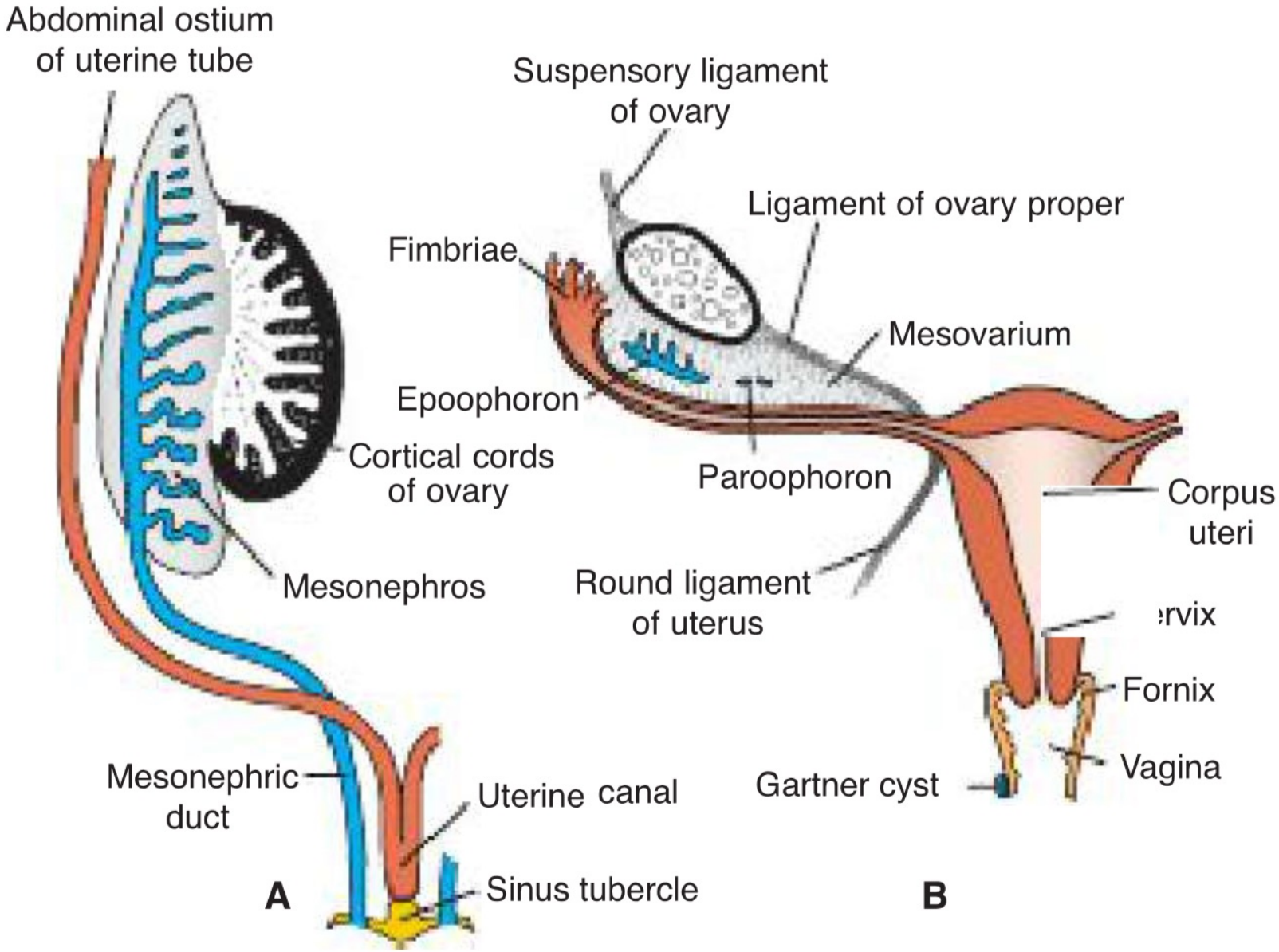
Differentiation of the ducts - females



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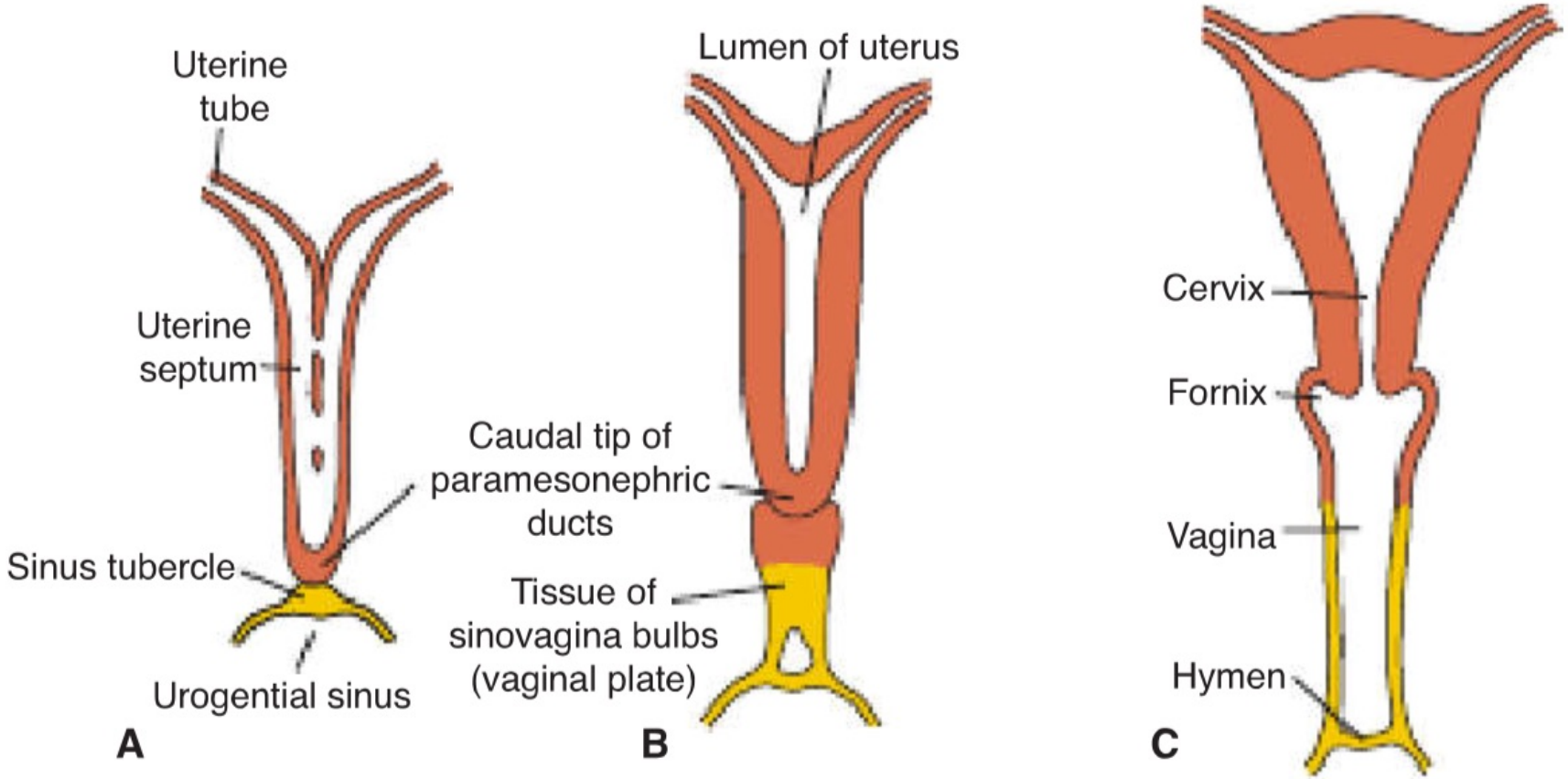
Development of the vagina



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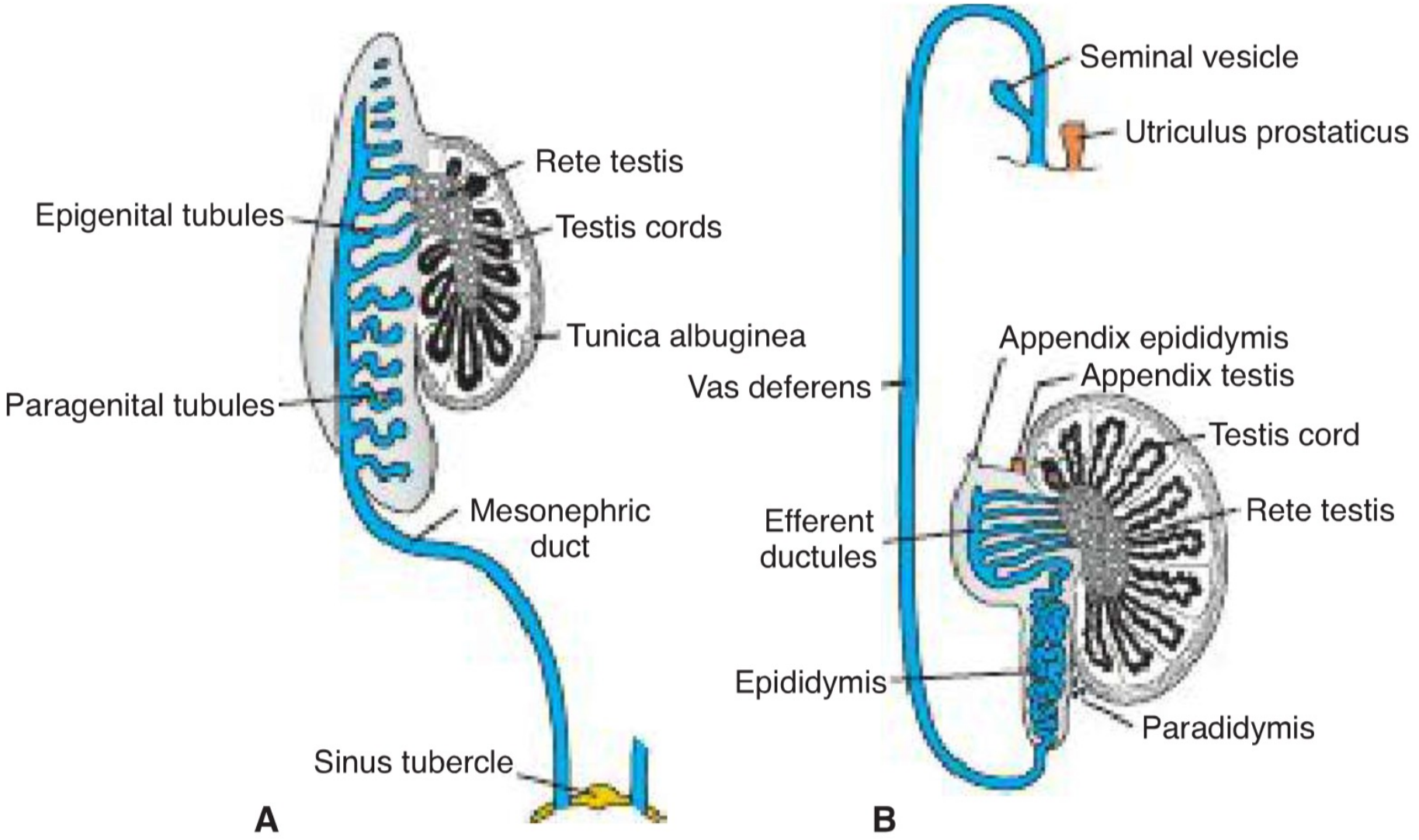
Differentiation of the ducts - males



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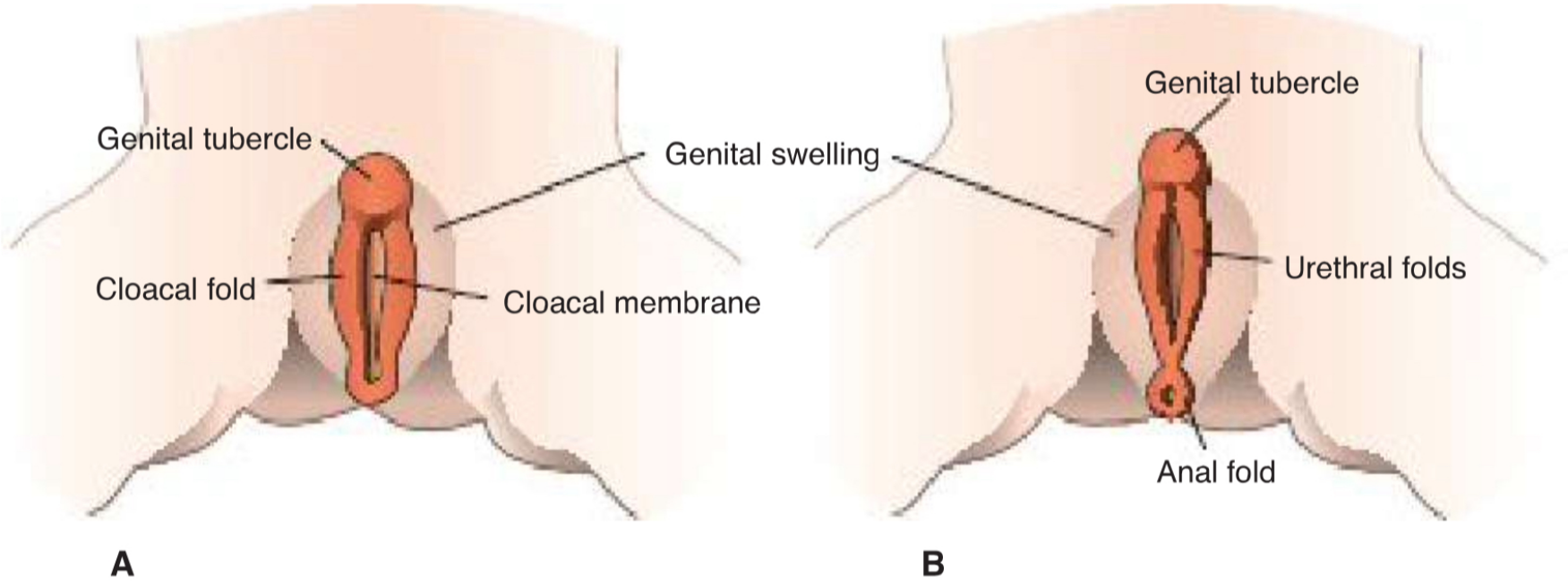
Indifferent stage of the external genitalia



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Disorders of sexual development (DSDs)

- Ambigous genitalia: clitoral hypertrophy/small penis with hypospadia
- 46, XX DSDs:
- - congenital adrenal hyperplasia, most common cause – 60 % of all DSDs
- 46, XY DSDs:
- androgen insensitivity syndrom (AIS): complete (CAIS), mild (MAIS) or partial (PAIS)
- 5-a-reductase deficiency: testosteron convertion into dihydrotestosteron is impaired

Ovotesticular DSDs - the caryotype is 46, XX in 70 % of cases

Chromosomal abberations

- Klinefelter syndrom – 47, XXY (XXXY…) – 1/1000 males: decreased fertility, small testes, decreased testosteron levels, gynecomastia in app. 33%

- Gonadal dysgenesis – oocytes are absent:

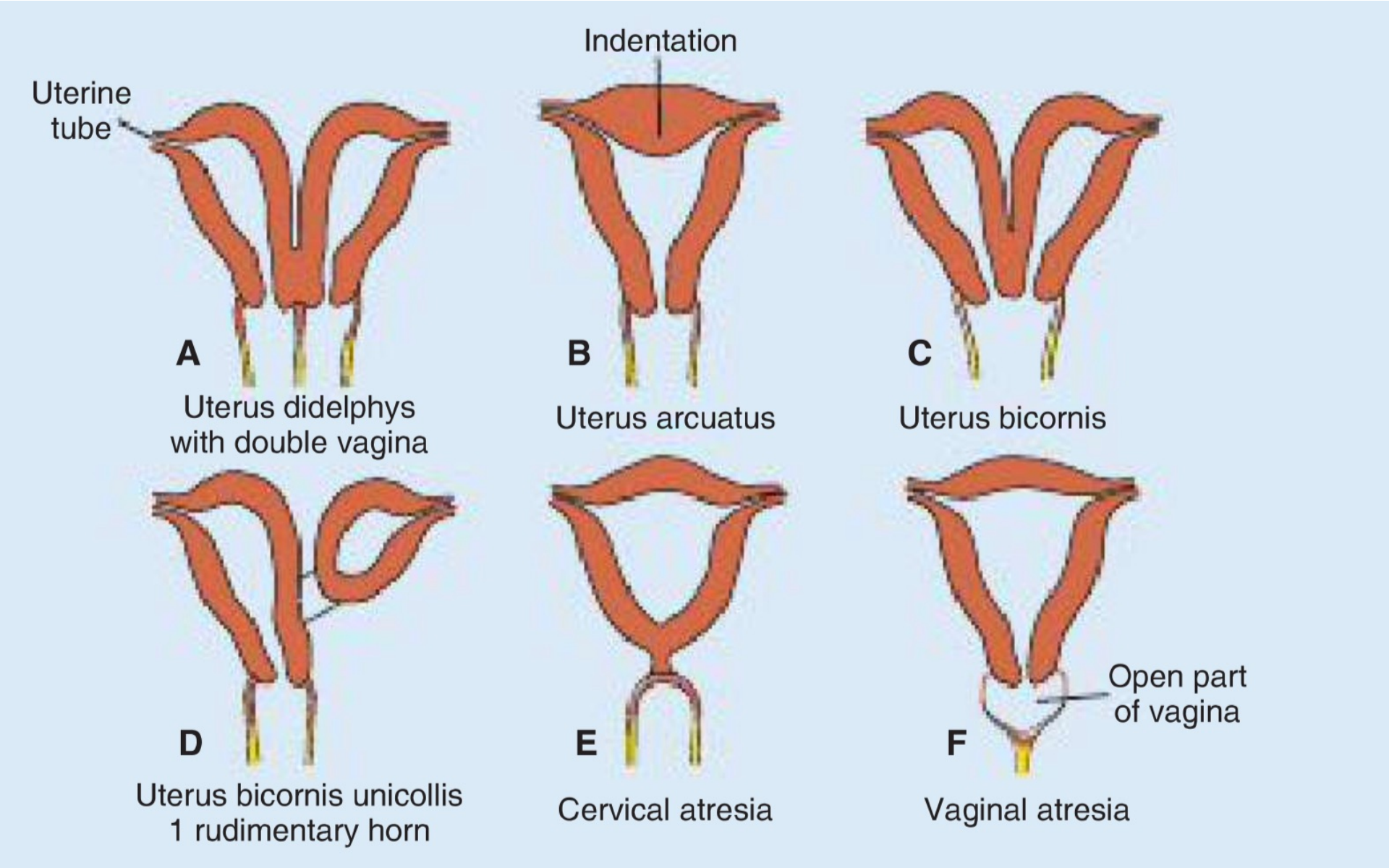
- Swyer syndrome – XY female gonadal dysgenesis – point mutation or deletions of the SRY gene
- Turner syndrome – 45, X

Hernias and cryptorchidism

- Congenital inderect inguinal hernia
- Hydrocele of the testis and/or spermatic cord

- Cryptorchidism

Uterine and vaginal defects



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Defects in male genitalia

- Hypospadia – 3-5/1000
- Epispadia – 1/30000 – most often associated with extrophy of the bladder and abnormal closure of the ventral body wall
- Micropenis – insufficient androgen stimulation – primary hypogonadism, hypothalamic or pituitary disfunction
- Bifid penis